

Search for

Limits History Details

Display

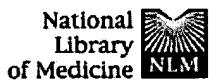
1: AAA79207. inositol polyphos...[gi:1019103]

[BLink](#), [Links](#)

LOCUS AAA79207 942 aa linear PRI 12-OCT-1995
 DEFINITION inositol polyphosphate 5-phosphatase.
 ACCESSION AAA79207
 VERSION AAA79207.1 GI:1019103
 DBSOURCE locus HUMINP5P accession M74161.1
 KEYWORDS .
 SOURCE Homo sapiens (human)
ORGANISM Homo sapiens
Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi;
Mammalia; Eutheria; Primates; Catarrhini; Hominidae; Homo.
 REFERENCE 1 (residues 1 to 942)
 AUTHORS Jefferson,A.B. and Majerus,P.W.
 TITLE Properties of type II inositol polyphosphate 5-phosphatase
 JOURNAL J. Biol. Chem. 270 (16), 9370-9377 (1995)
 MEDLINE 95238452
 PUBMED 7721860
 COMMENT Method: conceptual translation.
 FEATURES Location/Qualifiers
 source 1..942
 /organism="Homo sapiens"
 /db_xref="taxon:9606"
Protein 1..942
 /product="inositol polyphosphate 5-phosphatase"
CDS 1..942
 /gene="5ptase"
 /coded_by="M74161.1:<1..2830"
 ORIGIN
 1 vtvpepgaae srapgcdssg gcvsagasm dqsvaiqetl aegeycviav qgvlgcdsr
 61 qsrllglvry rlehggqeha lflythrrma itgddvslsq ivpvsrdftl eevspdgely
 121 ilgsdvtvql dtaelslvfq lpfgsqtrmf lhevaracpg fudsatrdpef lwlsryrcae
 181 leleemptprg cnsalvtwpg yatiggggsn fdglrpngkg vpmdqssrgq dkpeslqprq
 241 nkskseitdm vrssttivsd kahilsmqkf glrdtivksh llqkeedyty iqnfrrffagt
 301 ynvngqspke clrlwlsgni qapdvycvgf qeldlskeaf ffhdtpkeee wfkavsegih
 361 pdakyakvkl irlvgimlll yvkqehaayi seveaetvgt gimgrmgnkg gvairfqfh
 421 tsicvvnshl aahieeyerr nqdykdicsr mqfcqpdpssl ppltisnhdv ilwlgdlnyr
 481 ieeldvekvk klieekdfqm lyaydqlkiq vaaktvfegf tegeltfqpt ykydtgsddw
 541 dtsekcrapa wcdrilwkgk nitqlsyqsh malktsdhkp vssvfdigvr vvndelyrkt
 601 leeivrsldk menanipsvs lskrefcfqn vkymqlkves ftihngqvpc hfefinkpde
 661 esyckqwlna npsrgfilpd sdveidlelf vnkttatkl sgedkiedil vlhldrgkdy
 721 flsvsgnylp scfgspihtl cymrepildl pletiseltl mpvwtgddgs qldspmeipk
 781 elwmmvdly rnavqqedlf qgpglrsefe hirdcldtgm idnlsasnhs vaealllfle
 841 slpepvicys tyhnklecsg nytaskqvis tlpifhknvf hylmaflrel lknsaknhld
 901 enilasifgs lllrnphq kldmtekkka qefihqflcn pl

//

Revised: July 5, 2002.



PubMed	Nucleotide	Protein	Genome	Structure	PopSet	Taxonomy	OMIM	Books
Search	PubMed	<input type="button" value="▼"/>	for			<input type="button" value="Go"/>	<input type="button" value="Clear"/>	
Limits Preview/Index History Clipboard Details								

<input type="button" value="Display"/>	<input type="button" value="Abstract"/>	<input type="button" value="▼"/>	Show:	20	<input type="button" value="▼"/>	<input type="button" value="Sort"/>	<input type="button" value="▼"/>	<input type="button" value="Send to"/>	<input type="button" value="File"/>	<input type="button" value="▼"/>
--	---	----------------------------------	-------	----	----------------------------------	-------------------------------------	----------------------------------	--	-------------------------------------	----------------------------------

Entrez
PubMed

□1: Hum Mol Genet 1995 Dec;4(12):2245-50

[Related Articles](#), [Links](#)

PubMed
Services

Lowe syndrome, a deficiency of phosphatidylinositol 4,5-bisphosphate 5-phosphatase in the Golgi apparatus.

Suchy SF, Olivos-Glander IM, Nussbaum RL.

Laboratory of Genetic Disease Research, National Center for Human Genome Research, National Institutes of Health, Bethesda, Maryland 20892, USA.

The oculocerebrorenal syndrome of Lowe (OCRL) is an X-linked disorder characterized by congenital cataracts, renal tubular dysfunction and neurological deficits. The gene responsible for this disorder, OCRL-1, has been cloned and mutations identified in patients. The gene product (ocr1-1) has extensive sequence homology to a 75 kDa inositol polyphosphate 5-phosphatase. We report here that OCRL patients' fibroblasts show no abnormality in inositol polyphosphate 5-phosphatase activity, but are deficient in a phosphatidylinositol 4,5-bisphosphate [PtdIns(4,5)P₂] 5-phosphatase activity localized to the Golgi apparatus. Direct biochemical diagnosis of this human disease should now be possible. PtdIns(4,5)P₂ has been implicated in Golgi vesicular transport through its role in the regulation of ADP-ribosylation factor, phospholipase D and actin assembly in the cytoskeleton. The regulation of PtdIns(4,5)P₂ levels by PtdIns(4,5)P₂ 5-phosphatase may, therefore, be important in the modulation of Golgi vesicular transport. Given that the primary defect in OCRL is a deficiency of a Golgi PtdIns(4,5)P₂ phosphatase, we hypothesize that the disorder results from dysregulation of Golgi function and in this way causes developmental defects in the lens and abnormal renal and neurological function.

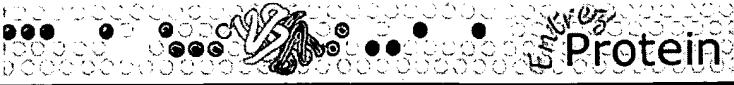
PMID: 8634694 [PubMed - indexed for MEDLINE]

<input type="button" value="Display"/>	<input type="button" value="Abstract"/>	<input type="button" value="▼"/>	Show:	20	<input type="button" value="▼"/>	<input type="button" value="Sort"/>	<input type="button" value="▼"/>	<input type="button" value="Send to"/>	<input type="button" value="File"/>	<input type="button" value="▼"/>
--	---	----------------------------------	-------	----	----------------------------------	-------------------------------------	----------------------------------	--	-------------------------------------	----------------------------------

[Write to the Help Desk](#)

[NCBI](#) | [NLM](#) | [NIH](#)

[Department of Health & Human Services](#)
[Freedom of Information Act](#) | [Disclaimer](#)



NCBI

PubMed Nucleotide Protein Genome Structure PopSet Taxonomy OMIM Books

Search for

Limits Preview/Index History Clipboard Details

Display

1: AAB03216. phosphatidylinosi...[gi:1399105]

[BLink](#), [Links](#)

LOCUS AAB03216 397 aa linear PRI 29-JUN-1996
 DEFINITION phosphatidylinositol (4,5)bisphosphate 5-phosphatase homolog.
 ACCESSION AAB03216
 VERSION AAB03216.1 GI:1399105
 DBSOURCE locus HSU45975 accession U45975.1
 KEYWORDS .
 SOURCE Homo sapiens (human)
 ORGANISM Homo sapiens
 Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi;
 Mammalia; Eutheria; Primates; Catarrhini; Hominidae; Homo.
 REFERENCE 1 (residues 1 to 397)
 AUTHORS Nussbaum,R.L.
 TITLE Direct Submission
 JOURNAL Submitted (11-JAN-1996) Robert L. Nussbaum, NCHGR, NIH, 49 Convent
 Drive, Bethesda, MD 20892, USA
 COMMENT Method: conceptual translation.
 FEATURES Location/Qualifiers
 source 1..397
 /organism="Homo sapiens"
 /db_xref="taxon:9606"
 /tissue_type="brain"
 /dev_stage="neonatal infant"
 /note="derived using ESTs, GenBank Accession Number R13943
 and R15390"
Protein
 1..397
 /name="phosphatidylinositol (4,5)bisphosphate
 5-phosphatase homolog"
CDS
 1..397
 /coded_by="U45975.1:<1..1194"
 ORIGIN
 1 arglhfvkfa idsdqlhqlw ekdqlnmakn twpilkgfqe gplnfaptfk fdvgtnkydt
 61 sakkrkpawt drilwkvpak gggpspsgrk shrlqvtqhs yrshmeytv dhkpvaqfl
 121 lqfafrrddmp lvrlevadew vrpeqavvry rmetvfarsr ss dwiglyrvg frhckdyvay
 181 vwakhedvdg ntyqvtfsee slpkghgdfi lgyyshnhsi ligitepfqi slpsselass
 241 stdsssgtsse geddstlell apksrspspg kskrhrsrsp glarfpglal rpssrerrga
 301 srspspqsr rr lsrvapdrss ngssrgssee gpsglpgpwa fppavprslg llpalrletv
 361 dpgggswgp drealapnsl spspqghrgl eegglgp
 //

Revised: July 5, 2002.

[Disclaimer](#) | [Write to the Help Desk](#)
[NCBI](#) | [NLM](#) | [NIH](#)



Sequence Revision History

PubMed Nucleotide Protein Genome Structure PopSet Taxonomy OMIM Books

Find (*Accession, GI number or Fasta style SeqId*)

Entrez Protein

Sequence Revision History				
Query	GI	Version	Update Date	Status
1420920	13254464	2	Mar 8 2001 17:16	Live
1420920	13254464	2	Mar 8 2001 17:15	Dead
1420920	1420920	1	Jul 16 1996 0:12	Dead

SeqId gi|1420920 was first seen at NCBI on Jul 16 1996 0:12

Related
resources

Revised: July 5, 2002.



NCBI Protein

PubMed Nucleotide Protein Genome Structure PopSet Taxonomy OMIM Books

Search Protein for 1420920 Go Clear

Limits Preview/Index History Clipboard Details

Display Summary Show: 20 Send to File

Entrez Protein

□1: [AAB03839](#)

gb|AAB03839.1|[1420920]

The entry was deleted. See [revision history](#) for details.

Related
resources

Revised: July 5, 2002.

[Disclaimer](#) | [Write to the Help Desk](#)
[NCBI](#) | [NLM](#) | [NIH](#)